X-Plain™
Phenylketonuria or PKU
Reference Summary

Phenylketonuria, or PKU, is an inherited disease. It can cause mental retardation if not treated early and for life.

PKU is rare. Only 1 out of every 15,000 babies in the United States is born with it. PKU is more common in people whose ancestors were Native American or Northern European.

Babies are checked for PKU right when they are born. PKU is treated with a special diet. Without treatment, people with PKU become mentally retarded.

This reference summary explains what PKU is. It discusses its causes, symptoms, diagnosis, and treatment.

What Is PKU?

Our bodies need protein to grow, make new cells and repair tissues. Protein is made of 20 building blocks called amino acids. Think of amino acids as 20 different Lego® blocks. They go together to make different shapes.

Protein comes from meat, fish, milk, eggs, beans and nuts that we eat. The body breaks protein down into amino acids. Any amino acids that are not needed are broken down and disposed of in the urine.

One amino acid is called phenylalanine. People with PKU cannot process phenylalanine. If phenylalanine is not processed, it builds up in the blood. This buildup affects nerve cells, causing brain damage and mental retardation.

Symptoms

Babies born with PKU act like any other baby for the first few months. If the PKU is not treated, they start losing interest in things when they are 3 to 6 months old.

When kids with untreated PKU are 1 year old, it is obvious they are not developing like they should. Children with untreated PKU have nerve and brain damage.

Untreated PKU sometimes makes the child smell musty. This is because the buildup of phenylalanine is in their breath, urine and sweat. They may also have dry skin, rashes or convulsions.

Kids with untreated PKU often have a small head called microcephaly.

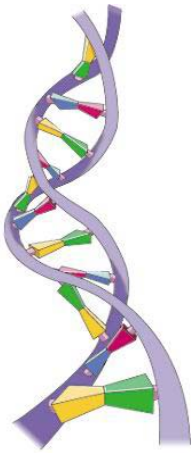
The body processes phenylalanine into tyrosine which produces melanin. Melanin causes darker skin and eyes. The body of kids with PKU cannot process phenylalanine into tyrosine. That is why kids with untreated PKU usually have very light skin and blue eyes.

Luckily, problems from PKU are rare. Most babies born with PKU are diagnosed and treated early.

Causes

PKU is inherited, which means children get it from their parents. However, even if parents do not have PKU, their children could still have it.

Genes tell the body to build certain substances. Every person has thousands of genes. Our genes make us look the way we do. They also have something to do with our health.



Each person has 23 pairs of chromosomes. Genes are found on the chromosomes. The gene linked to PKU is on chromosome 12.

The gene on chromosome 12 makes PAH protein. PAH is what helps the body process phenylalanine into tyrosine.

The PAH gene either works correctly or it is defective. We will call the good PAH gene that works correctly “P”. We will call the defective PAH gene that does not work correctly “p”.

Everybody has 2 PAH genes: 1 from their father + 1 from their mother. If both PAH genes are defective p genes, the person has PKU.

The good P gene is stronger than the defective p gene. Therefore, if a person has at least 1 P gene, they do not have symptoms of PKU.

- A PP person does not have PKU at all.
- A Pp person does not have PKU but carries the defective p gene. This is a PKU carrier.
- A pp person has PKU.

When a couple has a baby, each parent gives 1 gene that is like their own for each trait the child will have. Which gene the baby gets from each parent is random.

If both parents are PP, their child will not have PKU. Each parent can give only one PAH gene.

If both parents are pp, their child will definitely have PKU. Each parent can give only one p gene.

If one parent is PP and the other is Pp, their child has a 50% chance of being a Pp. Since a Pp is only a PKU carrier, they will never have a child that has symptoms of PKU. The PP parent will always give a good P gene.

If both parents are Pp, their child has a 1 in 4 chance of having PKU. In 2 out of the remaining 3 chances, the child will be a carrier.

It is important to remember that each time this couple has a baby, there is the same chance of the baby having PKU. If a couple’s first baby has a 25% chance (or 1 in 4) of having PKU, any other babies they have will have the same chance.

PKU Test

All U.S. states and territories test babies for PKU before they leave the hospital.

A nurse pricks the baby's heel and gets a few drops of blood. The blood is tested for PKU, as well as other genetic diseases.

The PKU test is very accurate if it is done when the baby is more than 24 hours old.

It is becoming common for mothers and babies to leave the hospital soon after birth. This makes it necessary to do the test before the baby is 24 hours old. If this happens, the doctor usually recommends that the baby be tested again when it is 1 to 2 weeks old.

Babies born at home or somewhere besides a hospital need to be tested for PKU and other diseases.

Treatment

Mental retardation can be prevented if a baby with PKU is started on treatment within the first 10 days of life. Treatment is a special diet that is low in phenylalanine and continues for the life of the child.

The goal of PKU treatment is to keep the level of phenylalanine between 2 and 6 mg/dl. The baby needs some phenylalanine to grow normally.

At first, the baby gets a special formula that contains protein but no phenylalanine. The baby can only have a little bit of breast milk or regular infant formula. A special dietician helps parents know how much the baby can have.

As the baby grows, it can start eating solid foods at the same age as other children. The only difference is the food must be very low in phenylalanine.

Later, the child can have certain vegetables, fruits and grain products that are low in phenylalanine. However, regular milk, cheese, eggs, meat, fish and other high protein foods are not allowed.

Since protein is necessary to grow and develop, the child still needs to drink the special formula. It is high in protein but has little or no phenylalanine.

People with PKU cannot have diet drinks or foods that contain aspartame. Aspartame is an artificial sweetener that has phenylalanine in it. It is usually called NutraSweet® or Equal®.

People with PKU should continue being seen at a medical center or clinic that specializes in PKU. The diet for each person with PKU is different. It depends on how much phenylalanine they can tolerate, their age, weight and other factors.

Anyone with PKU needs their blood tested regularly for phenylalanine levels. The diet is adjusted according to blood levels. Babies with PKU need to be tested frequently for the first year of life. During childhood, it may not be as often.

Individuals with PKU must stay on a special diet all through childhood and adolescence.

It is recommended to stay on the special diet for life.

Maternal PKU

There are around 3,000 young women with successfully treated PKU in the U.S. Most of them stopped their special diet when they were children because at that time, doctors believed it was safe.

Since these young women eat a normal diet, their phenylalanine levels are high. If they were to get pregnant, high levels of phenylalanine in the mother are very bad for the fetus.

In up to 90% of cases where a pregnant mother has high levels of phenylalanine, the baby is mentally retarded. Sometimes the baby also has a small head, heart defects, low birth weight and characteristic facial features.

The PKU diet after birth cannot help brain damage caused by the mother's high phenylalanine levels during pregnancy. Brain damage due to maternal PKU CANNOT be reversed after birth!

Brain damage caused by high phenylalanine levels in the mother CAN be prevented. The mother has to get back on the special diet at least 3 months before getting pregnant. She also has to stay on it during pregnancy.

With phenylalanine levels under control, women with PKU can have a healthy baby. The pregnant mother needs weekly blood tests to make sure phenylalanine levels are not too high. If they are high, we know that the fetus's levels are usually.

Summary

Phenylketonuria, or PKU, is an inherited disorder that can cause mental retardation if not treated early and for life.

Fortunately, testing allows babies with PKU to be diagnosed. Treatment consists of a special diet, which allows people with PKU to develop normally.

Parents of children with PKU and adults with PKU should discuss diet and treatment questions with a doctor at a special clinic for PKU.

Recent research holds some promise for people with PKU. Some researchers are trying to find ways to give people with PKU the PAH enzyme so they can use phenylalanine. Other researchers are trying to replace the PAH genes with ones that work so that phenylalanine does not build-up and cause nerve damage.